Diagnosis of endocardial cushion defect with cross-sectional and M-mode scanning echocardiography
Differentiation from secundum atrial septal defect[1]

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Twelve cases of endocardial cushion defect were studied before and after operation with ultrasono-cardio-tomography (tomography) (cross-sectional echocardiography, two-dimensional echocardiography, B-scan echocardiography) and M-mode scan along a horizontal section of the heart. For comparison, 20 healthy subjects, 18 cases of mitral valvular disease, 4 cases of congestive cardiomyopathy, 1 case of partial anomalous pulmonary venous drainage, and 25 cases of atrial septal defect of secundum type were also examined with the same technique.

In cases without cardiac malformation, the echo of the anterior mitral valve was usually continuous medially with that of the interatrial septum in the horizontal plane at the level of the membranous septum. This feature was clearly recorded in all cases with right heart enlargement.

In ostium secundum atrial septal defect the echo of the anterior mitral valve continued into that of the interatrial septum. An echo interruption was shown, indicating the defect itself to be in the middle part of the interatrial septum.

In all the cases of endocardial cushion defect which we examined, discontinuity was shown between the echo of the anterior mitral valve and that of the interatrial septum. This discontinuity was interpreted as indicating the defect itself. The mitral valve ring echo was close to the basal end of that of the interventricular septum, possibly reflecting an abnormal attachment of the mitral valve. In all cases, after operation, the echo of the artificial interatrial septum was recorded, continuous with that of the anterior mitral valve.

The features of the echocardiographic sweep from the anterior mitral valve to the interatrial septum were thus different in the three groups. These echocardiographic differences are thought to correspond to the anatomical differences between the normal, atrial septal defect of secundum type, and endocardial cushion defect, and are essential features differentiating them from each other.

At present, the diagnosis of endocardial cushion defect is based on haemodynamic and angiographic studies (Baron *et al.*, 1964). Conventional non-invasive examinations such as chest *x*-ray films, phonocardiography, and electrocardiography, help towards a diagnosis of this malformation. Incomplete right bundle-branch block with left axis deviation in the electrocardiogram is one of the characteristic features (Burchell, DuShane, and Brandenburg, 1960). However, this is not always found in this malformation (Toscano-Barbosa, Brandenburg, and Burchell, 1956) and occasionally it is seen in cases of atrial septal defect of secundum type (Harrison and Morrow, 1963). Therefore, to differentiate endocardial cushion defect from other cardiac malformations, including atrial septal defect of secundum type, a further non-invasive technique is needed.

Recently, echocardiography has been widely used

in clinical cardiology, because it has the advantage of being non-invasive with no discomfort to the patient. It has been proved to be a useful aid in the diagnosis of congenital heart disease (Chung et al., 1973; Dillon et al., 1973; Gramiak et al., 1973; Lundström, 1973; Meyer and Kaplan, 1972; Nimura et al., 1974; Popp et al., 1974). In endocardial cushion defect, several characteristic patterns in standard echocardiograms have been reported (Gramiak et al., 1972; Pieroni, Homcy, and Freedom, 1975; Williams and Rudd, 1974).

Ultrasono - cardio - tomography (tomography) (cross-sectional echocardiography, two-dimensional echocardiography, B-scan echocardiography) allows the relations between intracardiac structures to be seen, because it corresponds precisely to the anatomy of the heart (Ebina et al., 1967; King, 1973; King, Steeg, and Ellis, 1973; Nimura et al., 1971, 1974; Matsumoto, 1973; Matsumoto et al., 1975; Tanaka et al., 1971). Sahn et al. (1974) attempted to diagnose endocardial cushion defect by means of two-dimensional echocardiography with multi-crystal scan; however, they could not detect the defect itself but only the abnormalities of the echo of the mitral apparatus.

As previously reported by Nimura et al. (1971), Matsumoto (1973), and Matsumoto et al. (1975), tomographic visualization of the defect of the interatrial septum was obtainable in patients with ostium secundum atrial septal defect. This study was aimed at examining the anatomical abnormalities associated with endocardial cushion defect by ultrasono-cardio-tomography and M-mode scan. In combination, these two techniques were able to detect the interatrial septal defect and the abnormal attachment of the anterior mitral valve. The different site of the atrial septal defect in endocardial cushion defect compared with that in atrial septal defect of secundum type was clearly recognized by tomographic visualization.

Subjects and methods

A commercially available ultrasonograph, an Aloka SSD-30B, was used with a 2·25 MHz, 10 mm-diameter transducer with a pulse repetition rate of 1500 Hz. This equipment allowed us to record both standard and cross-sectional echocardiographic recordings by switching modes of display. To obtain the tomogram, the transducer was attached to the end of a guide arm giving reference to its position and angle (Fig. 1). The echoes at multiple points were recorded by slow manual scanning. The equipment was connected to a device, an Aloka SCU-1F, which had delay and gate circuits triggered by the electrocardiogram. The duration of the display time was set at 30 milliseconds by the gate circuit, and the onset of this display time was
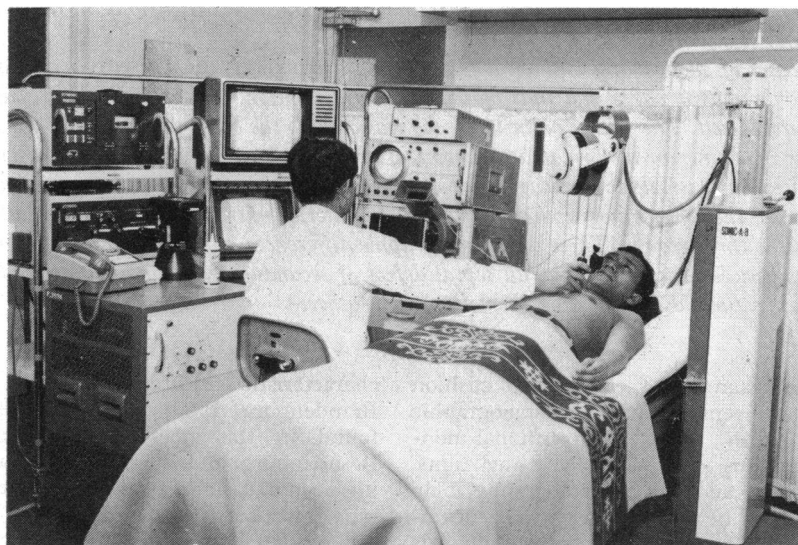

FIG. 1   *An echocardiographic examination using commercially available ultrasono-cardiotomograph. The transducer is attached to the end of the guide arm giving reference to its position and angle and rotated and moved slowly for scanning. The equipment is connected to a device with delay and gate circuits triggered by the electrocardiogram.*

set at a predetermined phase of a cardiac cycle by the delay circuit. This allowed us to record pictures at certain periods of the cardiac cycle, avoiding the blurring effect of the cardiac motion. It took about 100 beats to complete a tomographic record (Ebina *et al.,* 1967; King, 1973; Tanaka *et al.,* 1971).

The patients were examined in the supine position, during spontaneous respiration. The ultrasonic scans were along the horizontal section of the heart, with the transducer near the left sternal border in the third, fourth, and fifth intercostal space to obtain the tomogram and M-mode scan.
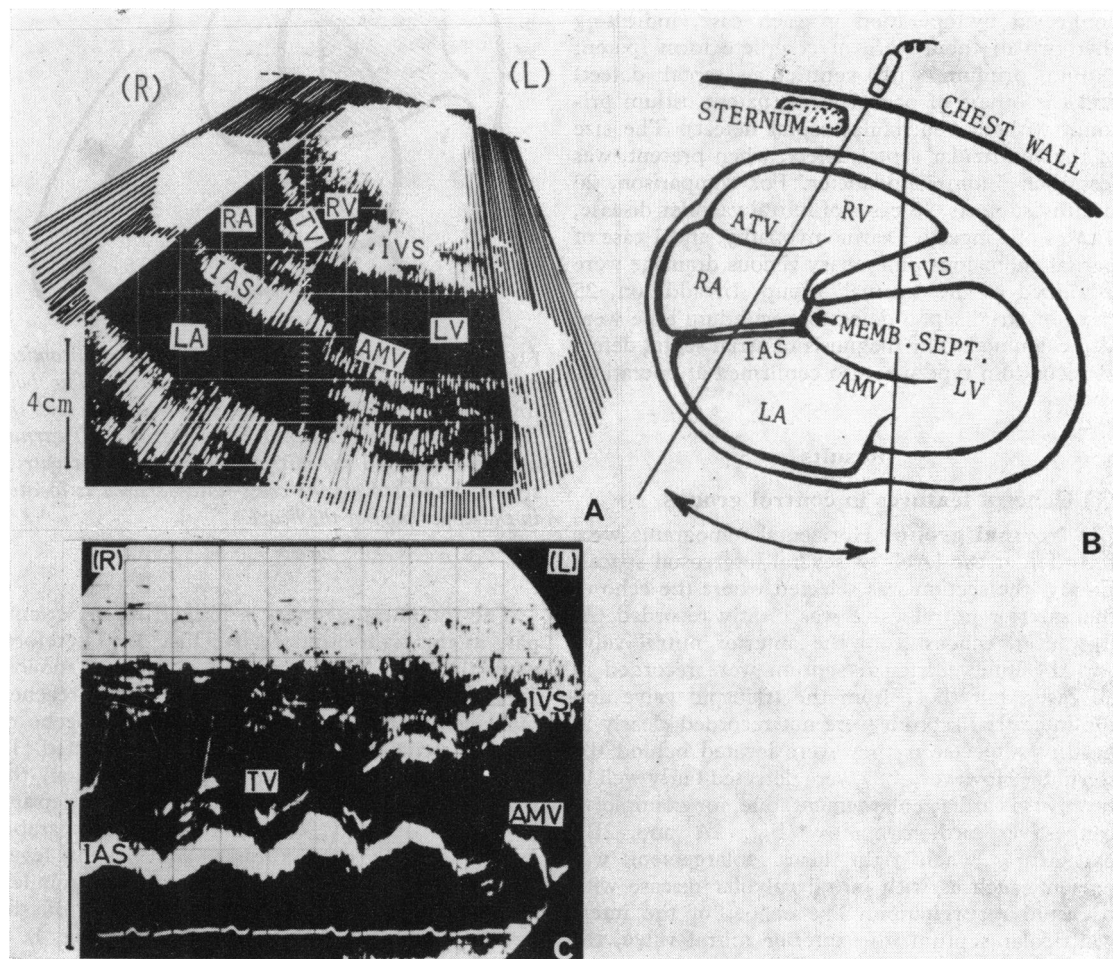

FIG. 2 *Horizontal section at the level of the membranous septum in the normal group. (A) A tomogram, viewed from the caudal side, in late systole. The interventricular septum, the interatrial septum, the tricuspid valve, and the anterior mitral valve were seen. The anterior mitral valve leaflet is continuous with that of the interatrial septum without interruption. The membranous septum, connecting the interventricular septum and the interatrial septum, was not clearly recorded. (B) A schematic drawing of the horizontal section of the heart. (C) An M-mode scan from the right to the left of the heart in the same plane as the tomogram. As observed in the tomogram, the interatrial septum connects with the anterior mitral valve leaflet behind the medial end of the interventricular septum. IVS=interventricular septum; IAS= interatrial septum; TV=tricuspid valve; AMV=anterior mitral valve; RV=right ventricle; RA=right atrium; LV=left ventricle; LA=left atrium; MEMB. SEPT.=membranous septum. (A 63-year-old woman with congestive cardiomyopathy.)*

Echoes on the tomogram were identified by reference to the motion patterns of corresponding echoes in the standard echocardiograms which were recorded with fixed beams in the area of the above-mentioned section.

Twelve patients with endocardial cushion defect ranging in age from 2 to 27 years were examined before and after operation. The diagnosis was confirmed by operation in each case, indicating that two of them were of complete form (patent ostium primum with ventricular septal defect) and the others of partial form (patent ostium primum without ventricular septal defect). The size of the ventricular septal defect, when present, was less than 5 mm in diameter. For comparison, 20 healthy subjects, 18 cases of mitral valvular disease, 4 cases of congestive cardiomyopathy, and 1 case of partial anomalous pulmonary venous drainage were examined as the 'normal' group. In addition, 25 cases of atrial septal defect of secundum type were also examined. The diagnosis of atrial septal defect of secundum type was also confirmed at operation.

Results

(1) General features in control groups

(A) Normal group Horizontal tomograms were recorded at the levels of several intercostal spaces. Firstly, the section was selected where the echo of the anterior mitral valve was clearly recorded. At this level, echoes from the anterior mitral valve and the interventricular septum were recorded in all cases, but those from the tricuspid valve and the interatrial septum were not recorded clearly in healthy subjects, as they were located behind the sternum. However, they were detected fairly well in cases with cardiac enlargement, due, for example, to congestive cardiomyopathy (Fig. 2A and 2B), particularly when right heart enlargement was present, such as with mitral valvular disease with tricuspid regurgitation. The echoes of the interventricular septum, the anterior mitral valve, the interatrial septum, and the tricuspid valve, running from left anterior, left posterior, right posterior, and right anterior, respectively, all seemed to approximate to one another. The echo of the membranous septum, which lies between the interventricular septum and the interatrial septum, was not usually recorded clearly. The echo of the interatrial septum was continuous with that of the anterior mitral valve in every case.

In the M-mode scan on the same section as the above tomogram, the continuity of the interatrial septum with the anterior mitral valve was also noted (Fig. 2C).
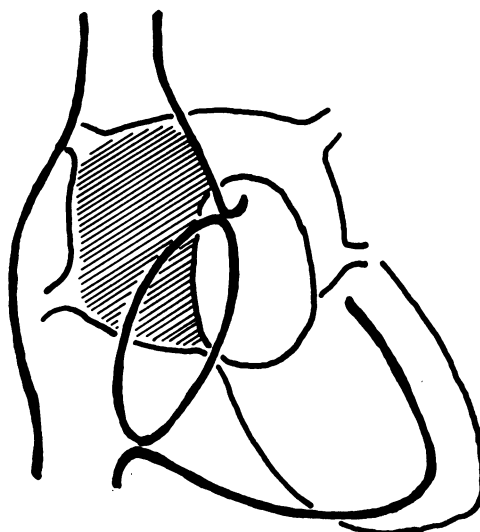

FIG. 3 *A schematic drawing of the anatomical relation between the right and left atria. The bold line indicates the right atrium and ventricle and the tricuspid annulus. The thin line shows the left atrium and ventricle and the mitral annulus. The interatrial septum indicated by the cross-hatched area is located in the upper part of the heart.*

The interatrial septum is located in the cranial part in the right atrial cavity (Fig. 3). Therefore, when the level of the horizontal section was too low (caudal), even if the ultrasound beam reached medially enough behind the sternum, the echo of the interatrial septum could not be detected. In other words, the lower the scanning section, the more posterior was the echo of the interatrial septum in the atrial cavity, eventually being inseparable from that of the posterior atrial wall. At this level, the echo of the interventricular septum ran from left anterior to right posterior joining that of the posterior wall with the mitral valve echo (Fig. 4).

If the level of the section was too high (cranial), the echo of the aorta was recorded, and the anterior mitral valve echo was continuous not with that of the atrial septum, but medially with that of the posterior wall of the aorta.

It was thus necessary to determine the level of the section for scanning carefully when examining cases of atrial septal defect of either secundum or primum type.

(B) Atrial septal defect of secundum type In the horizontal tomogram, the echo of the anterior mitral valve was continuous with that of the atrial
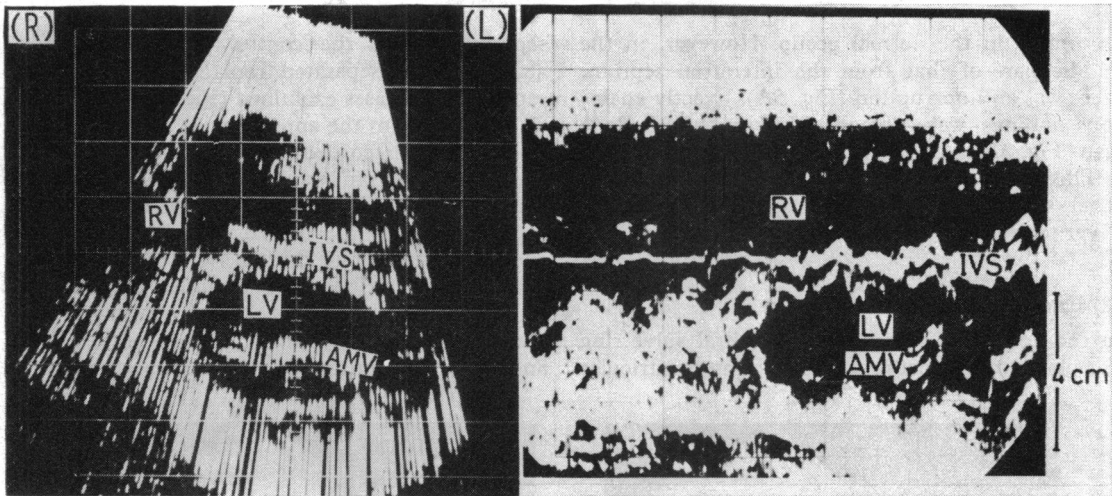
(A) **FIG. 4** *Horizontal echocardiograms at the lower (caudal) level in the normal group. (A) A* *(B)* *tomogram, viewed from caudal side, in late systole. The level of the section is almost at the mid portion of the left ventricle. The anterior mitral valve echo did not connect with the interatrial septum but with the posterior left ventricular wall along with that of the interventricular septum. (B) : An M-mode scan at the same level as in (A). The interventricular septum emerged from the posterior left ventricular wall. These observations indicate that the echocardiographic recording at this level does not allow visualization of continuity between the interatrial septum and the anterior mitral valve. (A 14-year-old boy with partial anomalous pulmonary venous drainage.)*
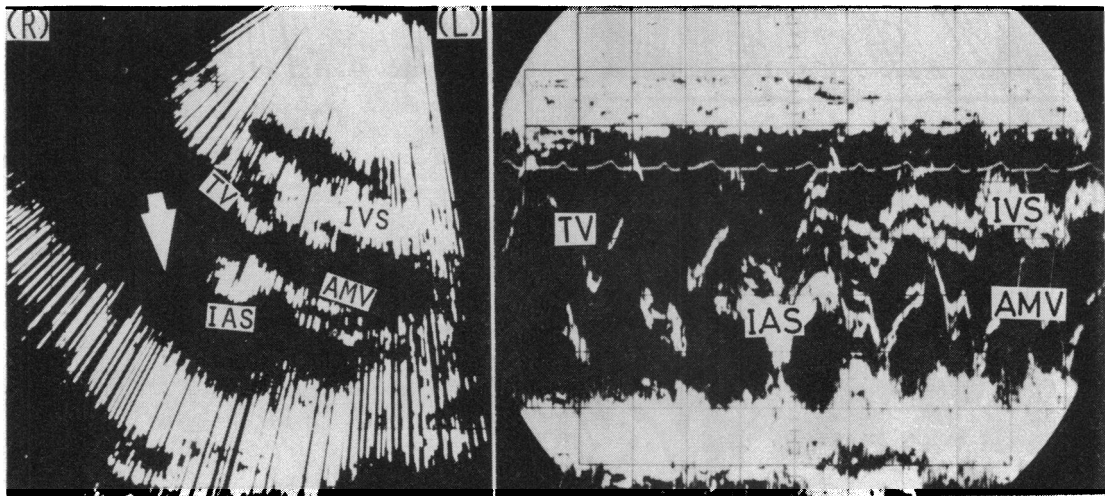
(A) **FIG. 5** *Horizontal echocardiograms at the level of the membranous septum in atrial septal* *(B)* *defect of secundum type. (A) A tomogram, viewed from caudal side, in late systole. The anterior mitral valve was connected to the interatrial septum behind the medial end of the interventricular septum. The interatrial septum was abruptly interrupted in the middle of the atrial area (indicated by the white arrow). (B) An M-mode scan from the right to the left. The echo of the interatrial septum discontinued in the middle of the atrial area. Before complete appearance, it was recorded only during diastole, probably because the ultrasound beam grazed the rim of the defect during diastole, but not during systole, because of the movement of the heart. The connexion between the interatrial septum and the anterior mitral valve was normal. (A 22-year-old woman.)*

septum as in the normal group. However, in the middle part of that from the interatrial septum, there was an interruption (Fig. 5A). Exactly analogous findings were demonstrable on the M-mode scan (Fig. 5B).

This interruption was detected in all 25 cases of atrial septal defect of secundum type, but was never found in the normal group.

(2) Endocardial cushion defect

In the horizontal tomogram, the mitral valve ring was closer to the medial end of the interventricular

septum than in the control groups, and ended abruptly, being separated from that of the interatrial septum in all cases examined (Fig. 6A and 6B and 7A). The echo of the anterior tricuspid valve leaflet was distinctly recorded and its relation to the interventricular septum was the same as that of the control groups. The tip of the tricuspid valve echo approximated to the medial end of the interventricular septum echo during systole (Fig. 6A) and separated from it during diastole (Fig. 6B). In three cases, the echo of the interatrial septum could be shown to arise from the atrial posterior wall and to be discontinuous with that of the anterior
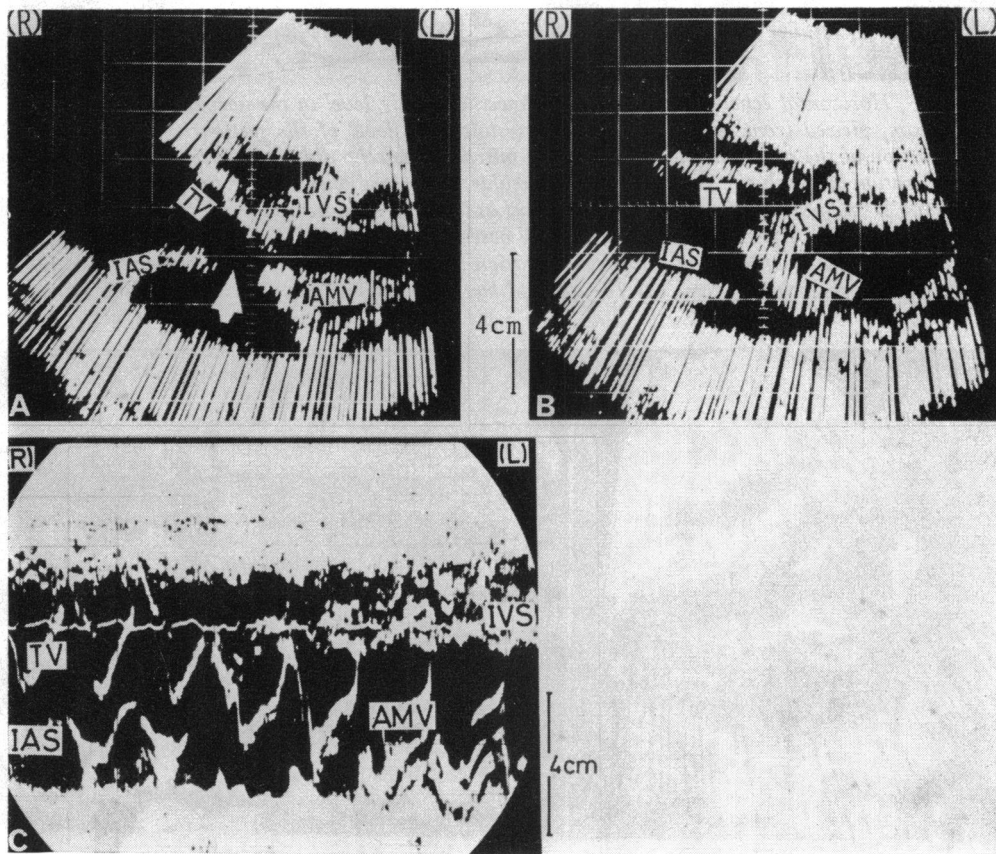

FIG. 6 *Horizontal echocardiograms at the level of the membranous septum in endocardial cushion defect. (A) A tomogram at late systole. The base of the anterior mitral valve leaflet was close to the medial end of the interventricular septum. An interruption (indicated by the white arrow) was visualized between echoes from the interatrial septum and the anterior mitral valve leaflet. (B) A tomogram at mid diastole. The mitral and tricuspid valves were in the open position. The tip of the tricuspid valve is separated from the medial end of the interventricular septum. (C) An M-mode scan from the right to the left. The interatrial septum abruptly disappeared in the atrial area, and there was no continuity between the interatrial septum and the anterior mitral valve. (A 15-year-old girl with partial form of endocardial cushion defect.)*
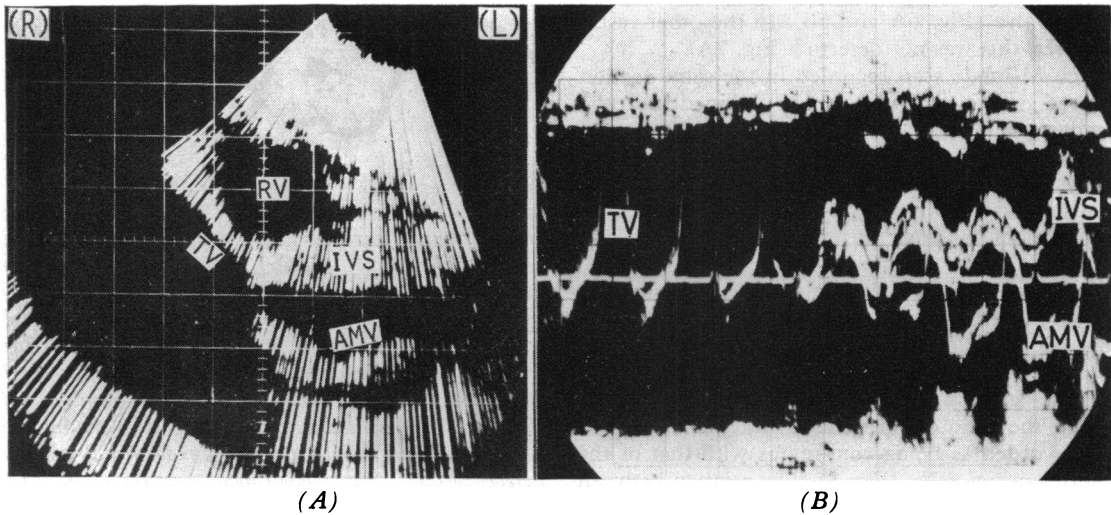
(A)   (B)

FIG. 7 *Horizontal echocardiograms at the level of the membranous septum in endocardial cushion defect (another example). (A) A tomogram at late systole. The annular end of the anterior mitral valve was recorded close to the medial end of the interventricular septum and was not connected to the interatrial septum, as in Fig. 6. However, the interatrial septum was not recorded. (B) An M-mode scan from the right to the left. The interatrial septum was not detected. The other findings were the same as those in Fig. 6. (A 27-year-old woman with the partial form of endocardial cushion defect.)*
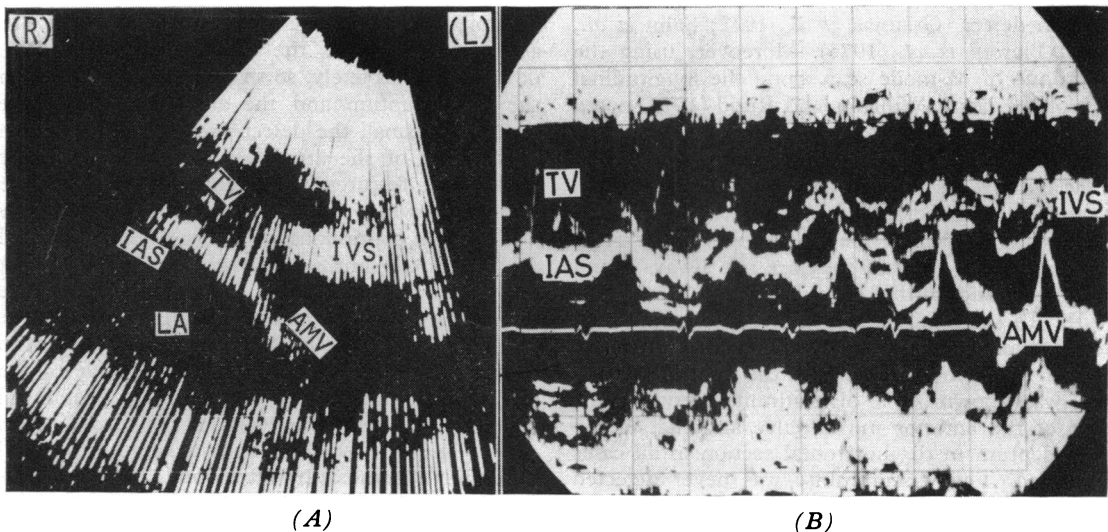

(A)   (B)

FIG. 8 *Horizontal echocardiograms in endocardial cushion defect after surgery. (A) A tomogram viewed from caudal side. (B) An M-mode scan from the right to the left. These echocardiograms revealed the artificial interatrial septum connecting to the anterior mitral valve without interruption. (A 22-year-old woman with the partial form of endocardial cushion defect.)*

mitral valve (Fig. 6A and 6B). In the other cases, however, this was not detected (Fig. 7A).

The M-mode scan recorded in the same section showed similar findings to those obtained from the tomograms. The echo of the anterior mitral valve apparently emerged from that of the interventricular septum (Fig. 6C and 7B). The echo of the anterior tricuspid valve often appeared to be continuous with that of the anterior mitral valve (Fig. 6C and 7B). In the same 3 patients as in the tomograms the echo of the interatrial septum was recorded as being separate from the anterior mitral valve (Fig. 6C), while in the remainder, this was not detected (Fig. 7B).

After surgical repair of the interatrial septal defect, the echo of the artificial interatrial septum was recorded as being continuous with that of the anterior mitral valve (Fig. 8). The relation between the anterior mitral valve and the interventricular septum was the same as that preoperatively in all cases examined (Fig. 8).

Discussion

Exaggerated anterior mitral valve excursion traversing the interventricular septum, prolonged mitral-septal apposition, a narrow left ventricular outflow tract, paradoxical movement of the interventricular septum, and increased right ventricular dimension have recently been reported as characteristic echocardiographic features of endocardial cushion defect (Gramiak *et al.*, 1972; Sahn *et al.*, 1974; Pieroni *et al.*, 1975). Moreover, using the technique of M-mode scan along the longitudinal axis of the heart, Williams and Rudd (1974) noted that the echo of the anterior mitral valve emerged from that of the anterior border of the left ventricular outflow tract. Each of these findings, which were also obtained in our study, is not sufficient on its own to confirm the diagnosis of endocardial cushion defect, because they are not always found in cases of endocardial cushion defect (Sahn *et al.*, 1974). In order to diagnose endocardial cushion defect, therefore, the site of the atrial septal defect should be demonstrated. In the present study, this was shown as a discontinuity between the echo of the anterior mitral valve and that of the atrial septum in the horizontal section in all cases examined. This discontinuity was never detected in patients with other types of heart disease including those with atrial septal defect of secundum type, and it directly reflected the anatomical basis of the atrial septal defect of endocardial cushion defect (Fig. 9).

It is important in clinical practice to differentiate between endocardial cushion defect and secundum

NORMAL   ASD   ECD   COMMON A-V CANAL

FIG. 9 *Schematic drawings of horizontal section of the normal heart, atrial septal defect of secundum type, endocardial cushion defect, and common atrioventricular canal. The membranous septum (*) connects the interventricular septum, the septal tricuspid valve, the interatrial septum, and the anterior mitral valve. In the case of atrial septal defect of secundum type (ASD), the membranous septum is normal, and there is a defect in the middle part of the interatrial septum. In endocardial cushion defect (ECD), the membranous septum is not developed. The septal leaflet of the tricuspid valve and the anterior mitral valve leaflet are directly attached to the interventricular septum, so that there is no continuity between the interatrial septum and the anterior mitral valve leaflet. In common atrioventricular canal (COMMON A-V CANAL), an extreme form of endocardial cushion defect, the septal leaflet of the tricuspid valve and the anterior part of the anterior mitral valve are fused together to form a single leaflet, which is not connected to the interatrial septum.*

type of atrial septal defect. In the case of secundum atrial septal defect the endocardial cushion is developed completely, so that the relation between the atrial septum and the anterior mitral valve leaflet is normal, the defect itself being located in the middle of the atrial septum (Fig. 9). These anatomical features were demonstrated by the tomogram, which showed the anterior mitral valve leaflet to be continuous with that of the interatrial septum in the horizontal plane, while the defect itself was represented by an interruption in the middle part of the atrial septum (Fig. 5). Thus, the difference in the site of the interruption of the echo in the scan from the anterior mitral valve to the interatrial septum corresponded to the anatomical difference between the two types of atrial septal defect, and its demonstration makes the essential differential diagnosis by ultrasound. In our laboratory, we have never failed in a diagnosis of endocardial cushion defect, either false positive or false negative, using these criteria.

Another malformation in endocardial cushion defect is abnormal attachment of the anterior mitral valve. The anterior mitral valve does not attach to the membranous septum, but to the interventricular

septum directly, because the endocardial cushion is undeveloped (Fig. 9). In the present study, the echo of the base of the anterior mitral valve leaflet was recorded close to that of the medial end of the interventricular septum in all cases of endocardial cushion defect in comparison with that in the control groups, and this relation was not altered by corrective surgery. This finding corresponds to the abnormal attachment of the anterior mitral valve. In the control groups, on the other hand, the base of the anterior mitral valve leaflet was some distance from the medial end of the interventricular septum, the gap corresponding to the membranous septum.

Another feature of endocardial cushion defect is a cleft mitral valve. Sahn *et al.* (1974) presented multiple echoes in the mitral area on the echocardiogram and Pieroni *et al.* (1975) showed multiple representation of the systolic image of the mitral valve as an indication of the mitral cleft. In the present study, such echocardiographic findings were obtained in some cases with mitral cleft, which were confirmed later by surgery. However, these findings were not considered to represent features which were specific to the mitral cleft since repair of the cleft with 'teflon' or pericardium did not always eliminate these abnormal echoes, and since a similar echocardiographic pattern was obtained in cases of mitral valve disease or even congestive cardiomyopathy (personal observation). It is considered that further examination is necessary to differentiate one from the other.

The difference between complete and incomplete forms, i.e. the presence of the defect of the interventricular septum, was not detected in the present study. The defect of the interventricular septum in our cases was less than 5 mm in diameter, so that it was probably not detected on account of insufficient resolution of the equipment. In the case of an extreme form of endocardial cushion defect (common atrioventricular canal), the septal tricuspid valve and the anterior part of the anterior mitral valve are fused together to form one large leaflet (Fig. 9). Of cases examined in the present study, the M-mode scan along the horizontal section gave an apparent continuity between the anterior tricuspid valve and the anterior mitral valve, erroneously suggesting that two valves were fused together even in cases of partial form. However, the tomogram revealed that the anterior tricuspid valve was separate anteriorly from the interventricular septum during diastole (Fig. 6B), and displayed the individual movement of those two valves. Therefore, such continuity on the M-mode scan does not always indicate the common atrioventri-

cular valve. This apparent continuity might be caused by the direct attachment of the anterior mitral valve to the interventricular septum, since the anatomical relation between the anterior tricuspid valve and the interventricular septum was normal. Careful interpretation should be made when the M-mode scan gives apparent continuity or discontinuity between echoes, as discussed recently by French and Popp (1975) in another context.

In summary, horizontal echocardiographic searching at the level of the membranous septum is useful in the diagnosis of endocardial cushion defect and in the differentiation of this malformation from atrial septal defect of secundum type. The definitive diagnosis of endocardial cushion defect can be made when the echocardiogram reveals the discontinuity between the anterior mitral valve and the interatrial septum.

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